Evaluation and Management of Cleft Lip and Palate

A DEVELOPMENTAL PERSPECTIVE

SECOND EDITION

David J. Zajac, PhD, CCC-SLP, FASHA
Linda D. Vallino-Napoli, PhD, CCC-SLP/A, FASHA

Plural Publishing Inc.

PLURAL PUBLISHING
INC.

9177 Aero Drive, Suite B
San Diego, CA 92123

email: information@pluralpublishing.com
website: https://www.pluralpublishing.com

Typeset in 10.5/13 Palatino by Flanagan's Publishing Services, Inc.
Printed in the United States of America by Bradford & Bigelow

Library of Congress Cataloging-in-Publication Data:
Names: Zajac, David J., author. | Vallino-Napoli, Linda D., author.
Title: Evaluation and management of cleft lip and palate : a developmental perspective / David J. Zajac, Linda D. Vallino-Napoli.
Description: Second edition. | San Diego, CA : Plural Publishing, Inc., [2026] | Includes bibliographical references and index.
Identifiers: LCCN 2024031270 (print) | LCCN 2024031271 (ebook) | ISBN 9781635505351 (paperback) | ISBN 9781635504446 (ebook)
Subjects: MESH: Cleft Lip | Cleft Palate | Velopharyngeal Insufficiency | Speech Disorders—prevention & control
Classification: LCC RD525 (print) | LCC RD525 (ebook) | NLM WV 440 | DDC 617.5/22—dc23/eng/20240708
LC record available at https://lccn.loc.gov/2024031270
LC ebook record available at https://lccn.loc.gov/2024031271

Contents

Preface

We are pleased to provide a new edition of our text. Since publication of the first edition, there have been changing clinical perspectives and new research on key topics in the evaluation and care of children and adults with craniofacial anomalies. We cover these new developments and alert the reader to their clinical importance.

As with the first edition, we intend this book to be a concise, practical, and evidenced-based text on cleft lip and palate and related craniofacial disorders for advanced undergraduate students, graduate students, and professionals in speech-language pathology. Students and professionals in related disciplines such as dentistry, medicine, psychology, and social work also may find this book useful in providing information on individuals with craniofacial conditions. Indeed, we have been delighted to hear from colleagues in dentistry who have enthusiastically endorsed the text.

Cleft palate with or without cleft lip is a congenital defect that varies both in its severity and impact on facial/oral structures and communication. Treatment of individuals is typically a long and often-complicated process—extending into early adulthood and beyond—that is best accomplished in a team setting. In addition to the speech-language pathologist, other team members typically include a plastic surgeon, dentist, orthodontist, oral and maxillofacial surgeon, geneticist, social worker, and psychologist. Although all members of the team contribute to the habilitation of the individual throughout his/her lifespan, certain team members assume critical roles at specific times of life. The purpose of this book is to provide the student and/or professional in speech-language pathology and other healthcare professions the information needed to (a) evaluate and treat communication disorders associated with cleft palate regardless of their primary place of employment (i.e., craniofacial team, hospital, school, or private practice), and (b) understand the complex—and sometimes controversial—surgical and dental management of individuals.

To help achieve these goals, the material in the book is presented in a developmental framework that emphasizes the most critical needs of the individual from birth to adulthood. This organizational approach has both practical and conceptual advantages. Practically, it allows the reader to access information more readily according to the age and presenting condition of the individual (i.e., birth, lip repair, palate repair, alveolar bone grafting, maxillary advancement). Conceptually, it chronicles the lifelong impact of craniofacial birth defects on the individual and elucidates the timing and rationale of surgical, dental, and behavioral interventions.

Part I provides necessary fundamentals for the student and/or professional. Chapter 1 reviews embryology, anatomy, and physiology of the facial, oral, and velopharyngeal structures that are affected by clefts. Chapter 2 describes the types and causes of clefts with an emphasis on embryological classification. The controversy regarding timing of palate repair is discussed from the perspective of speech and language development. Chapter 3 provides an overview of genetics and the

terminology used to categorize and identify congenital anomalies. A select group of craniofacial anomalies most likely to be encountered by the professional is presented.

Part II focuses on evaluation and management of the individual from birth to three years of age. Chapter 4 describes normal feeding physiology, feeding problems associated with cleft palate, and approaches to facilitate feeding prior to palate repair. Chapter 5 describes presurgical and surgical management of cleft lip and palate. Chapter 6 covers the almost universal occurrence of otitis media with effusion, hearing loss that can occur in infants with cleft palate and current methods of management. Chapter 7 reviews early linguistic development in infants with cleft palate and intervention strategies before and after palate repair.

Part III focuses on evaluation and management of the individual from age three throughout the middle school years. Chapter 8 describes the resonance, nasal emission, articulation, voice, fluency, and intelligibility characteristics of children with repaired cleft palate. Chapter 9 provides detailed coverage of perceptual assessment and an overview of instrumental assessment techniques. Chapter 10 describes practical approaches to treating children with articulation problems in the school setting. Chapter 11 presents an overview of behavioral, surgical, and prosthetic options to manage velopharyngeal inadequacy that persists following initial palate surgery. Chapter 12 describes the orthodontic preparation and surgical correction of clefts of the alveolus.

Part IV focuses on evaluation and management of adolescents and adults with cleft palate. Chapter 13 describes the rationale and timing of maxillary advancement in adolescents to improve facial esthetics, dental occlusion, and articulation. Chapter 14 discusses the adult with cleft palate including persistent concerns, quality of life, and transition to adult-focused care.

Although clefts of the lip and/or palate are among the most frequently occurring birth defects, the actual number of individuals affected in the United States is relatively low. It is not unusual, therefore, for speech-language pathologists working in the schools to seldom encounter children with clefts. It is our hope that the materials in this book will be a valuable resource for clinicians. Certain content that is typically included in other texts has been omitted in this book. In order to maintain conciseness and adhere to a developmental structure, we have chosen not to include separate chapters on the functioning of craniofacial teams and the psychosocial aspects of individuals with craniofacial anomalies. Nevertheless, we have integrated relevant information on these subjects within different chapters throughout the book. It is evident that these areas hold significant importance. Maternal reactions to an infant born with a cleft are covered in Chapter 4, for example, and learning disabilities of children with cleft palate are reviewed in Chapter 10.

Finally, the book has been written with a goal to cite evidence-based sources to support evaluation and intervention approaches. In some areas, there is little objective evidence available to guide clinical decision-making. In those areas, we note the lack of evidence and suggest directions for future research. It is our hope that this book will not only inform but also challenge clinicians in speech-language pathology and other health professions to provide the best evidence-based evaluation and management of individuals with cleft palate and craniofacial anomalies as possible.

Contributors

Brenda Louw, D.Phil. SLP
ASHA Fellow
Professor Emerita
East Tennessee State University
Johnson City, Tennessee
Chapter 14

Joseph A. Napoli, MD, DDS
Professor of Clinical Surgery
Perelman School of Medicine, University of Pennsylvania.
Division of Plastic, Reconstructive, and Oral Surgery
Children's Hospital of Philadelphia
Philadelphia, Pennsylvania
Chapters 11, 12, and 13

Jamie L. Perry, PhD, CCC-SLP
ASHA Fellow
Professor and Chair
Department of Communication Sciences and Disorders
East Carolina University
Greenville, North Carolina
Chapter 1

Dennis M. Ruscello, PhD, CCC-SLP
ASHA Honors, ASHA Fellow
Professor Emeritus of Communication Sciences and Disorders
West Virginia University
Morgantown, West Virginia
Chapter 10

Nancy J. Scherer, PhD, CCC-SLP
ASHA Fellow
Professor and Chair
Department of Speech and Hearing Sciences
Arizona State University
Tempe, Arizona
Chapter 7

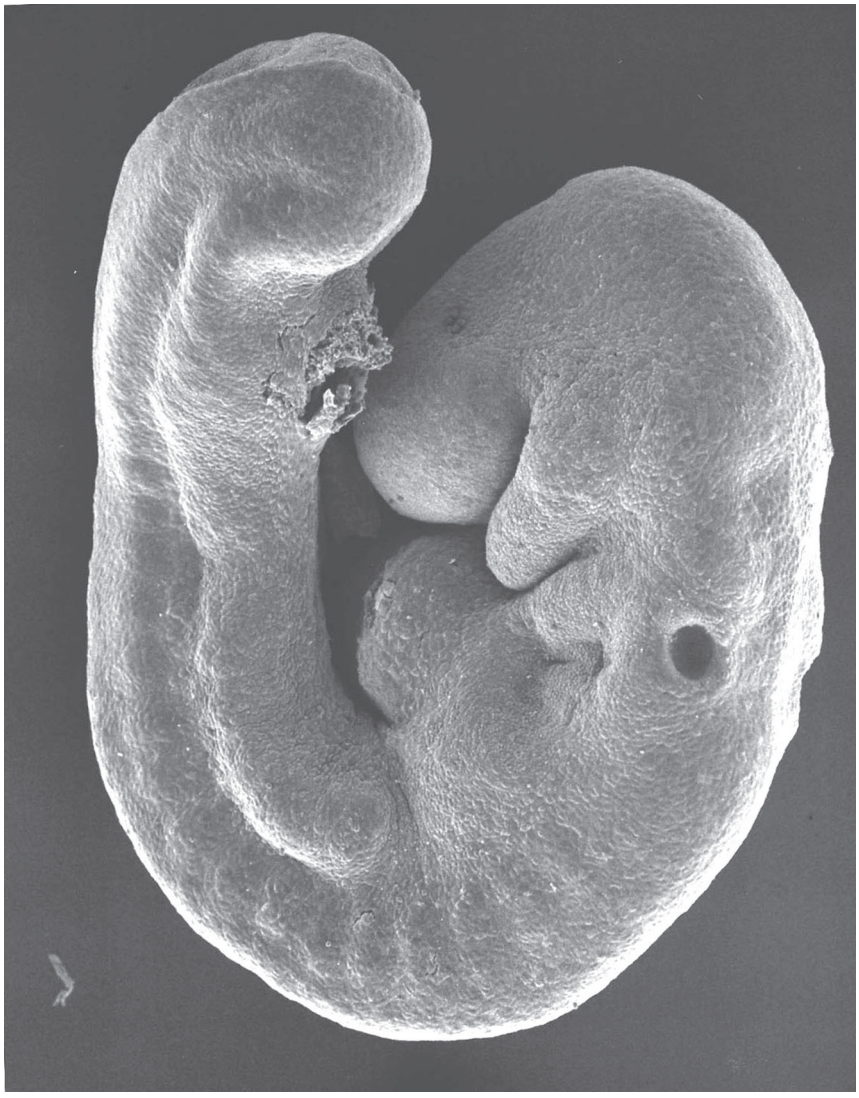
Linda D. Vallino-Napoli, PhD, CCC-SLP/A
ASHA Fellow
Senior Research Scientist
Head of the Craniofacial Outcomes Research Laboratory
Center for Pediatric Auditory and Speech Sciences
Nemours Children's Health
Wilmington, Delaware
Clinical Professor of Pediatrics
Sidney Kimmel Medical College–Thomas Jefferson University
Philadelphia, Pennsylvania
Adjunct Professor, Department of Communication Sciences and Disorders
University of Delaware
Newark, Delaware
Chapters 13 and 14

Natalie R. Wombacher, PhD, CCC-SLP
Department of Speech and Hearing Sciences
Arizona State University
Tempe, Arizona
Chapter 7

David J. Zajac, PhD, CCC-SLP
ASHA Fellow
Craniofacial Center
Professor, Department of Craniofacial and Surgical Care
Adams School of Dentistry
University of North Carolina at Chapel Hill
Chapel Hill, North Carolina
Chapters 1 and 11

PART I

Fundamentals



We intend this book to serve as a roadmap to the diagnosis and care of individuals with orofacial clefts and other craniofacial conditions from birth through adulthood. To do so, the speech-language pathologist (SLP) and other health care providers must know certain fundamentals. Chapter 1, "Orofacial and Velopharyngeal Structure and Function," provides information on embryology, normal anatomy, and physiology. Major landmarks of the face, nose, and oral cavity are identified and detailed descriptions of the velopharyngeal muscles and functions are provided. Chapter 2, "Clefts of the Lip and Palate," describes the types of clefts that are commonly encountered in the clinic. Special attention is given to submucous clefts as these may be subtle, difficult to identify, and may not have functional consequences. Causes of clefts and epidemiology relative to prevalence and recurrence are reviewed. Finally, Chapter 3, "Genetics and Craniofacial Anomalies," provides an overview of genetics and the terminology used to categorize and identify congenital anomalies. A select group of craniofacial anomalies that impact communication is presented. The information in Part I sets the stage for the remainder of the book.

1

Orofacial and Velopharyngeal Structure and Function

Jamie Perry and David J. Zajac

INTRODUCTION

Orofacial clefts can involve structural anomalies of the upper lip and gum ridge, nose, hard palate, and soft palate to various degrees. If a cleft is part of a syndrome or sequence, then additional craniofacial anomalies may be present involving the lower jaw, face, ears, and skull. To understand the nature and management of clefts—including the types, impact on feeding, hearing and speech, and surgical repair—health care providers must have a fundamental understanding of orofacial and velopharyngeal structures and function. The purpose of this chapter is to review (a) embryologic development of the face, (b) structures of the face, nasal cavity, oral cavity, pharynx, and velopharynx, and (c) velopharyngeal function of normal speech production.

EMBRYOLOGY OF THE FACE

To facilitate an understanding of the types and occurrence of oral clefts, a basic knowledge of normal embryologic development is essential. As reviewed below, the face largely develops from the first and second pharyngeal arches beginning at approximately the 4th week of gestation.

Formation of the Primary Palate—Upper Lip and Premaxilla

At approximately the 2nd week of human gestation, the embryo has a flat, disc-shaped appearance and consists of two cell layers, the *epiblast* and the *hypoblast*. During a process called gastrulation, cells of the epiblast begin to invaginate or fold in at the caudal (tail) end of the embryo. This process leads to the formation of three distinct cell layers: the *ectoderm* (previously the epiblast), the *mesoderm* (formed from the epiblast), and the *endoderm* (previously the hypoblast). These three cell layers will continue to grow and eventually form all of the specialized cells and organs of the fetus. Ectodermal cells will develop into neural and surface ectoderm that will compose the central

nervous system and outer covering of the body, respectively. Mesodermal cells will contribute to the formation of bone and connective tissue, and endodermal cells will contribute to the development of the digestive system.

The rapid folding and growth of the embryo changes its appearance from a flat disc to a more human-like form by the end of the 4th week. At this time, the embryo has developed into a C-shaped form with its cranial and caudal ends tightly curved (Figure 1–1). The head and neck of the embryo at this stage comprise approximately half its length. Also at this stage, the developing forebrain, *first* and *second pharyngeal arches*, the inner ear, heart, and upper limb are clearly discernable. The forebrain and pharyngeal arches develop from neural crest cells that migrate from the dorsal (back) side of the embryo to the front. The first pharyngeal arch has both *maxillary* and *mandibular* prominences that will eventually develop into the upper and lower jaws. The second pharyngeal arch will contribute to formation of the

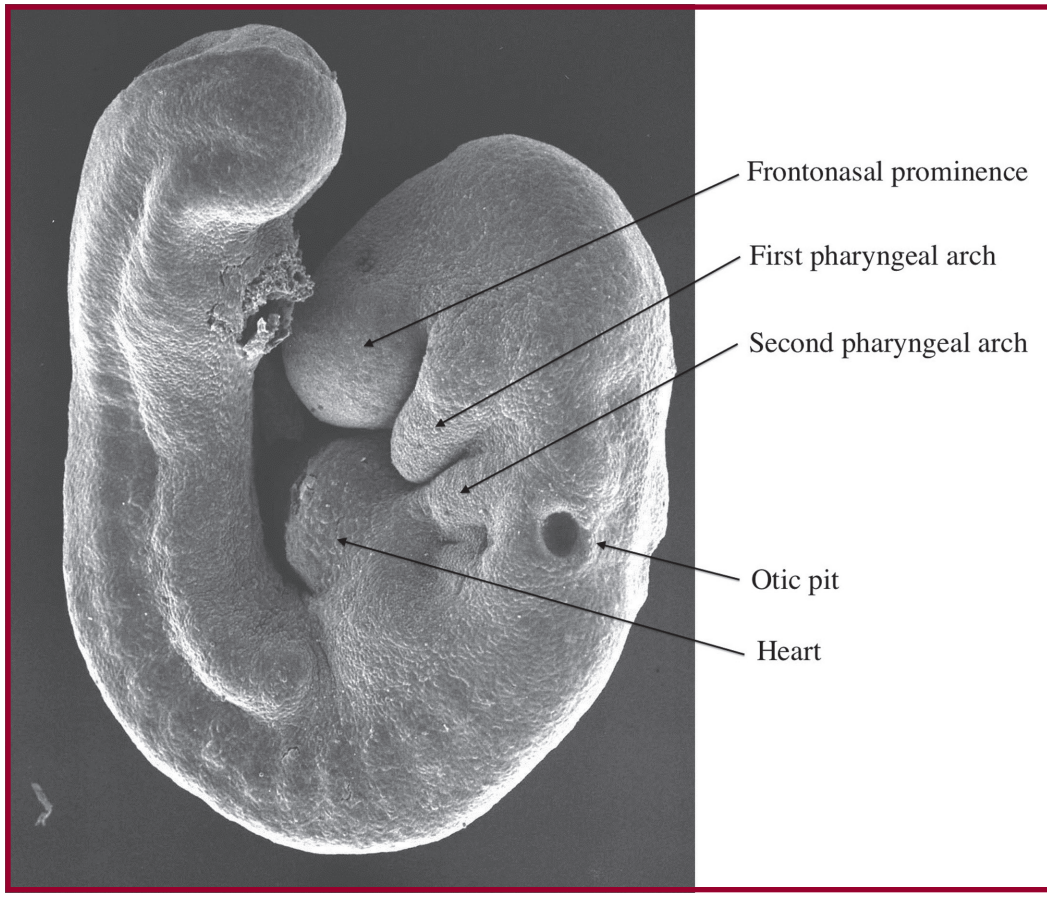


Figure 1–1. Scanning electron micrograph of mouse embryo at Gestation Day 9 (human age approximately 27 days). (Courtesy of Kathleen K. Sulik, PhD, Department of Cell Biology and Physiology, University of North Carolina at Chapel Hill.)

hyoid bone and ear. These structures, the first and second pharyngeal arches, are the primary contributors to the development of the face. The first pharyngeal arch will provide tissue prominences that develop into the *primary palate*, which consists of the premaxilla and the central portion of the upper lip.

Figure 1–2 shows development of the head of the embryo during the 5th week of gestation. Two nasal pits (indentions) are formed by migration of neural crest cells from the forebrain that develop *medial* and *lateral* tissue prominences. The nasal pits are lined with neural ectodermal cells called the olfactory placodes. This tissue will eventually form the olfactory nerve. Below the nasal pits is an opening that will develop into the *mouth (stomodeum)*. During the 6th week of gestation, the upper lip is formed by union of the medial and lateral nasal prominences with each other and with the *lateral maxillary* prominence, tissue that developed from the first pharyngeal arch. The medial nasal prominences continue to merge in the midline to complete formation of the upper lip and the philtral ridges (Figure 1–3). The medial nasal prominences continue to grow inward resulting in the *premaxilla*, a triangular section of bone that will contain the four maxillary incisors.

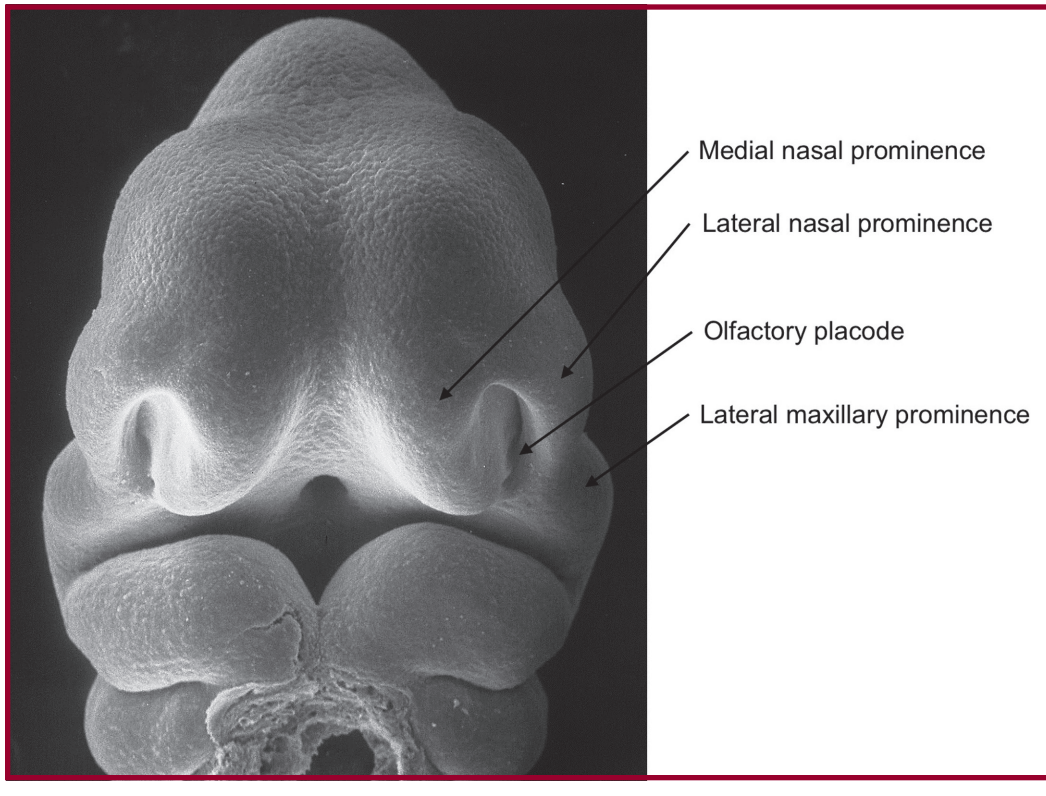


Figure 1–2. Scanning electron micrograph of mouse embryo at Gestation Day 10 (human age approximately 5th week). (Courtesy of Kathleen K. Sulik, PhD, Department of Cell Biology and Physiology, University of North Carolina at Chapel Hill.)

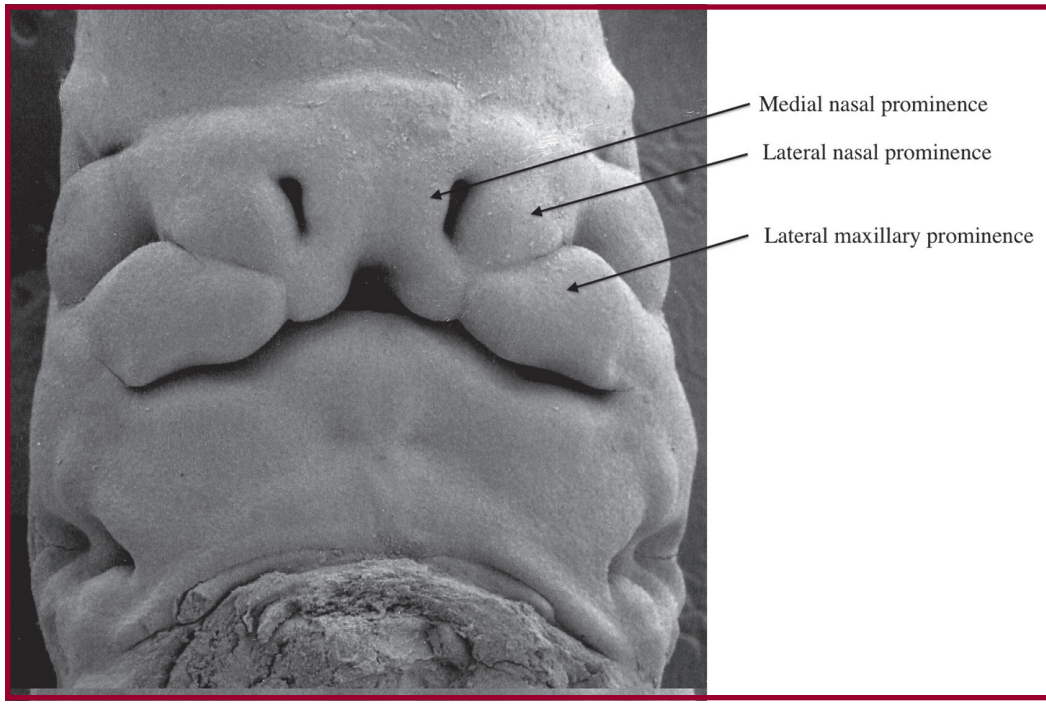


Figure 1–3. Scanning electron micrograph of human embryo at approximately 6 weeks' gestation. (Courtesy of Kathleen K. Sulik, PhD, Department of Cell Biology and Physiology, University of North Carolina at Chapel Hill.)

Formation of the Secondary Palate

The secondary palate begins to form after the primary palate at approximately 8 weeks' gestation. Initially, the paired palatal shelves are in a floppy, vertical position with the developing tongue interposed between the shelves (Figure 1–4). The palatal shelves are part of the lateral maxillary prominences that develop from the first pharyngeal arch. During the 9th week, the shelves begin to elevate and assume a horizontal position above the tongue before fusing. As illustrated in Figure 1–4, however, the tongue presents an obstacle to palatal shelf elevation and fusion. Some embryologists have proposed that the tightly curled embryo needs to grow in order to allow extension of the neck and forward growth of the mandible, resulting in lowering of the tongue prior to palatal elevation (Diewert, 1974; Zeiler et al., 1964). Others, however, have suggested that mandibular growth and tongue lowering actually occur following palatal fusion and that palatal fusion itself triggers tongue lowering and forward growth of the mandible (Burdi, 2006; Kjaer et al., 1993). It is also possible that mouth-opening reflexes are responsible for lowering the tongue prior to shelf elevation (Humphrey, 1969).

Regardless of the actual mechanism of tongue lowering, the palatal shelves elevate and begin to approximate each other in the midline (Figure 1–5). Fusion begins at the *incisive foramen* and continues in a posterior direction until completed